



RP2 gene

RP2, ARL3 GTPase activating protein

Normal Function

The *RP2* gene provides instructions for making a protein that is essential for normal vision. The RP2 protein is active in cells throughout the body, including cells that make up the light-sensitive tissue at the back of the eye (the retina). However, the function of the RP2 protein is not well understood. Studies suggest that it may be involved in transporting proteins within the retina's specialized light receptor cells (photoreceptors). Its role in other types of cells is unknown.

Health Conditions Related to Genetic Changes

retinitis pigmentosa

More than 70 mutations in the *RP2* gene have been identified in people with the X-linked form of retinitis pigmentosa. This condition primarily affects males, causing night blindness in early childhood followed by progressive daytime vision loss.

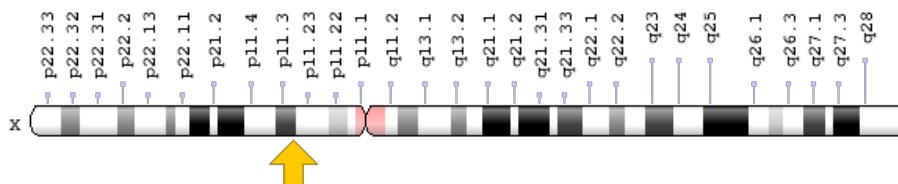
RP2 gene mutations account for 10 to 15 percent of all cases of X-linked retinitis pigmentosa.

Most mutations in the *RP2* gene lead to the production of an abnormally short version of the RP2 protein. A few mutations change single building blocks (amino acids) in the RP2 protein. These changes alter the structure and function of the protein, which probably disrupts the stability or maintenance of photoreceptor cells. A gradual loss of photoreceptors underlies the progressive vision loss characteristic of retinitis pigmentosa.

Chromosomal Location

Cytogenetic Location: Xp11.3, which is the short (p) arm of the X chromosome at position 11.3

Molecular Location: base pairs 46,836,912 to 46,882,358 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DELXp11.3
- KIAA0215
- NM23-H10
- NME10
- protein XRP2
- retinitis pigmentosa 2 (X-linked recessive)
- TBCCD2
- XRP2
- XRP2_HUMAN

Additional Information & Resources

GeneReviews

- Nonsyndromic Retinitis Pigmentosa Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1417>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28RP2%5BTIAB%5D%29+OR+%28retinitis+pigmentosa+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BA%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- RP2 GENE
<http://omim.org/entry/300757>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=RP2%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10274
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6102>
- RetNet: Summaries of Genes and Loci Causing Retinal Diseases
<https://sph.uth.edu/retnet/sum-dis.htm>
- UniProt
<http://www.uniprot.org/uniprot/O75695>

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